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Tuberous Sclerosis Complex: a Case Study

Rimma Gamirova [1,2,3]

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Abstract
The article is a contribution to the international database of tuberous sclerosis case studies. As tuberous sclerosis complex (Bourneville–Pringle disease) is a rare genetically determined polysystemic disease, each clinical case is very significant. The first clinical symptoms of the disease usually appear immediately after birth, but they differ in significant polymorphism and affect brain, different organs, and systems, including changes in the skin, nervous system, eyes, and inner organs. Accurate diagnosis of tuberous sclerosis is fundamental to proper medical supervision and treatment. The article describes modern genetic and clinical criteria of diagnosis in detail; the specificity of the clinical case and problems emerged during follow-up of the patient, diagnostic errors, and findings. The author proves the importance of a multidisciplinary approach not only in diagnosis but also in the treatment of tuberous sclerosis including neuropsychological approaches.

Keywords Tuberous sclerosis · Bourneville–Pringle disease · Genetic disease · Hamartoma · Epileptic seizures · Angiofibroma · Tuberous sclerosis complex

1 Introduction

Bourneville–Pringle disease, or Bourneville disease, or tuberous sclerosis (TS), or tuberous sclerosis complex (TSC), is a rare genetically determined polysystemic disease, the primary manifestations of which are benign tumors and lesions of the skin and various organs, including the brain [1–3]. Patients with tuberous sclerosis often suffer from epilepsy, mental retardation, and autistic disorders. The incidence of the disease is 1 per 10,000 newborns [4]. The development of tuberous sclerosis is determined by two genes localized in segment 34 of the long arm of the 9th chromosome (tuberous sclerosis, 1st type—TSC1, which codes for the hamartin protein, cytogenetic location is in 9q34.13) and in the segment 13 of the short arm of the 16th chromosome (tuberous sclerosis, 2nd type—TSC2, which codes for the tuberin protein, cytogenetic location is in 16p13.3) [5].

✉ Rimma Gamirova
r-gamirov@mail.ru

[1] Laboratory of Clinical Linguistics, Kazan Federal University, Tatarstan str., Kazan 420008, Russia

[2] Department of Pediatric Neurology, Kazan State Medical Academy, 11 Mushtari str., Kazan 420012, Russia

[3] Department of Fundamental Basis of Clinical Medicine, Kazan Federal University, 76 K.Marks str., Kazan 420012, Russia

It is necessary to underline that in 75% of cases, it is a spontaneous mutation; in 25%, it is a transmission by autosomal dominant type of inheritance from parents [6].

There are several case studies of Bourneville–Pringle disease (O.Chtourou et al., 2005; A.Cichoń-Mikołajczyk et al., 2006; S.Mimura et al., 2006; P.Dyrla et al., 2008; C.Sofoudis et al., 2016) [7–11] which have scientific and practical significance, and each new case should be described in order to create the effective paths of diagnosis, as the disease has different clinical manifestations (they may be associated with the skin, nervous system, eye bulbs, heart, kidneys, liver, and intestines, be presented in the form of developmental disorders).

2 Materials and Methods

1. *The diagnostic criteria* for tuberous sclerosis have not been updated for 15 years since the last Clinical Consensus in 1998, and only in 2012, they were critically analyzed and revised by the Conciliation Commission, consisting of 79 specialists from 14 countries. According to updated diagnostic criteria, the signs of tuberous sclerosis can be divided into the following:

   A. Genetic diagnostic criteria
      Identification of a genetic defect (mutation) of